General

Guideline Title

Updated, web-based nutrition management guideline for PKU: an evidence and consensus based approach.

Bibliographic Source(s)

Singh RH, Cunningham AC, Mofidi S, Douglas TD, Frazier DM, Hook DG, Jeffers L, McCune H, Moseley KD, Ogata B, Pendyal S, Skrabal J, Splett PL, Stembridge A, Wessel A, Rohr F. Updated, web-based nutrition management guideline for PKU: an evidence and consensus based approach. Mol Genet Metab. 2016 Jun;118(2):72-83. [118 references] PubMed

Guideline Status

This is the current release of the guideline.

This guideline meets NGC's 2013 (revised) inclusion criteria.

Recommendations

Major Recommendations

Definitions for the strength of the recommendations (Strong, Fair, Weak, Consensus, Insufficient Evidence) and need for clinical action (Imperative, Conditional) are provided at the end of the "Major Recommendations" field.

Nutrient Intake

Question

For individuals with phenylketonuria (PKU), what nutrient intakes are associated with positive outcomes?

Recommendations

Meet the individual's recommended phenylalanine (PHE) intake (for anabolism and maintaining an appropriate blood PHE concentration) by adjusting intact protein intake. (Fair, Imperative)

Provide a total protein intake (from a combination of intact protein and amino acid-based medical food) approximately 50% higher than the dietary reference intake (DRI) for infants and children from birth to 4 years of age and 20% to 40% higher than the DRI for those over 4 years of age. The amount of medical food prescribed is based on the difference between the total protein recommendation and the intact protein allowance. (Fair, Imperative)

Provide supplemental tyrosine (TYR) if blood TYR concentrations are consistently below the normal range. (Fair, Conditional)

With the exception of recommended intake for protein, PHE, and TYR, individuals with PKU should meet the same DRI for age- and gender-

specific nutrient/micronutrients and energy as healthy individuals in the general population. (Weak, Imperative)

Blood PHE Concentrations

Question

For individuals with PKU, what blood PHE concentrations are associated with positive outcomes?

Recommendations

Maintain lifelong blood PHE between 120 and 360 μmol/L. (Fair, Imperative)

Treatment should be initiated in individuals with PKU whose blood PHE exceeds 360 μmol/L. (Weak, Imperative)

Strategies for Nutrition Intervention

Question

For individuals with PKU, which nutrition interventions are associated with positive outcome?

Recommendations

Choose medical foods to meet recommended nutrient intake and achieve optimal adherence. When incomplete medical foods are chosen, ensure that vitamin, mineral, energy, and/or fat intake is supplemented from other sources when necessary. (Weak, Imperative)

Plan consumption of medical food throughout the day, in several well-spaced intervals, to allow optimal blood PHE concentrations and dietary PHE tolerance. (Strong, Imperative)

Encourage use of breast milk, when possible, either from direct breast feeding or use of expressed breast milk, as the source of PHE (and intact protein) in infants. (Fair, Conditional)

Gradually introduce solids, to replace the equivalent amount of PHE/intact protein in infant formula or breast milk, when the infant is developmentally ready (usually at 4–6 months of age). (Fair, Imperative)

Minimize elevation of blood PHE during illness by treating the underlying illness, meeting protein and energy needs, and preventing dehydration and electrolyte imbalance. (Consensus, Imperative)

Ensure appropriate PHE intake in individuals with PKU by having accurate data regarding PHE content of foods, and effective and convenient methods of planning and monitoring dietary PHE intake. (Weak, Imperative)

Encourage all individuals to follow treatment recommendations throughout their lives; including those who have relaxed their diet restrictions and those who have never been treated. Recognize and address individual barriers that may impede success. (Fair, Imperative)

Adopt clinic procedures that enhance adherence to the nutritional recommendations of "diet for life" by providing individualized educational strategies, referrals to appropriate social service and mental health professionals, age-appropriate group activities, and a plan for transition from pediatric to adult clinical services. (Fair, Imperative)

Monitoring Nutrition Intervention

Question

For individuals with PKU, monitoring of which parameters is associated with positive outcomes?

Recommendations

Monitor dietary records to assess adequacy of nutrient intake in supporting appropriate growth and nutritional status. If intake is suboptimal, modify individual dietary recommendations and counseling to improve adherence. (Strong, Imperative)

Monitor age-specific anthropometrics. (Fair, Imperative)

Routinely monitor clinical indicators and biochemical markers for deficiency or excess of nutrients whose intake may not be optimal in an individual on a PHE-restricted diet (PHE, TYR, protein, iron and vitamin D). (Strong, Imperative)

Monitor clinical indicators and biochemical markers when indicated by circumstances such as rapid growth, pregnancy, poor compliance with

management recommendations, or consumption of an incomplete medical food. (Fair, Conditional)

Monitor neurocognitive development. (Fair, Imperative)

Assess quality of life using age- and disorder-specific instruments when possible. (Weak, Conditional)

Nutrition Intervention with Alternative or Adjunctive Therapies

Question

For individuals with PKU whose therapy includes adjunctive therapy or other therapy options (sapropterin or large neutral amino acids [LNNA]), what dietary considerations are needed for positive outcomes?

Recommendations

When treatment with sapropterin (pregnancy class C) is appropriate, combine with diet therapy to improve blood PHE and/or clinical status, and develop individualized therapy plans to provide best outcome. (Strong, Conditional)

Conduct a PHE challenge to determine maximal dietary PHE tolerance when sapropterin response brings blood PHE to within control range, or to clarify a sapropterin response when historical blood PHE is already within control range. (Strong, Conditional)

Modify dietary therapy in individuals responsive to sapropterin to accommodate increased PHE tolerance. Liberalization should reflect increased PHE/intact protein intake, decreased medical food intake, and vitamin/mineral supplementation as appropriate. Monitor nutritional status and educate individuals regarding modified dietary recommendations. (Strong, Conditional)

Individualize and closely monitor sapropterin therapy when used in special populations, such as: infants and young children, pregnancy, and late- or untreated adults. (Weak, Conditional)

Consider LNAA supplementation in adults with PKU who are unable to achieve metabolic control with diet or other adjunctive therapy. LNAA therapy is not recommended for use in infants, young children, or women who are pregnant or may become pregnant. (Weak, Conditional)

When LNAA therapy is chosen, provide 20% to 30% of total protein intake from LNAA supplements, and the remaining 70% to 80% from intact dietary protein. Total protein intake should meet DRI requirements (0.8 g/kg/day). Monitor adequacy of protein intake and plasma amino acids to prevent essential amino acid deficiencies. (Weak, Conditional)

Nutrition Intervention Before, During, and After Pregnancy

Question

For women with PKU, what nutritional therapies are associated with positive outcomes during pregnancy planning, pregnancy, and the postpartum period (including lactation)?

Recommendations

Maintain blood PHE between 120 and 360 µmol/L before, during, and after pregnancy. (Strong, Imperative)

Monitor weight gain, dietary intake, and biochemical parameters to ensure nutrient adequacy and metabolic control during pregnancy. (Fair, Imperative)

Prescribe a diet that meets nutritional needs of pregnancy and promotes adequate weight gain. (Fair, Imperative)

Avoid LNAA monotherapy during pregnancy. (Consensus, Imperative)

Use of sapropterin should be evaluated during pregnancy on a case-by-case basis, and may be appropriate especially in women with moderate or mild forms of PKU who are not able to maintain blood PHE in the recommended treatment range for pregnancy. (Consensus, Imperative)

Facilitate access to psychosocial support as necessary to maintain dietary therapy in pregnancy. (Consensus, Conditional)

Encourage women with PKU to maintain dietary therapy after pregnancy and to breast-feed their infants. (Weak, Imperative)

Modify PKU therapy and collaborate with other care-givers to support nutritional and metabolic needs of women with multiple pregnancies, gestational diabetes, and other special circumstances. (Insufficient evidence, Conditional)

Definitions

Recommendation Ratings and Clinical Actions/Application

Strength of Evidence for Recommendation Ratings	
Strong	The benefits clearly exceed the harms (or the harms clearly exceed the benefits in the case of a strong negative recommendation); and the quality of the supporting evidence is good. In some clearly identified circumstances, strong recommendations may be made based on lesser evidence when high-quality evidence is impossible to obtain and the anticipated benefits strongly outweigh the harms.
Fair	The benefits exceed the harms (or the harms clearly exceed the benefits in the case of a negative recommendation), but the quality of evidence is not as strong as above. In some clearly identified circumstances, recommendations may be made based on lesser evidence when high-quality evidence is impossible to obtain and the anticipated benefits outweigh the harms.
Weak	The quality of evidence that exists is suspect or well-done studies show little clear advantage for one approach over another.
Consensus	Expert opinion (determined from consensus methodology) supports the recommendation even though the available scientific evidence did not present consistent results, or studies were lacking.
Insufficient Evidence	There is a lack of pertinent evidence (from research and clinical practice) and/or an unclear balance between benefits and harms.
Clinical Action/Application	
Imperative	The recommendation is broadly applicable to the target population without conditions.
Conditional	The recommendation clearly defines a specific situation that limits its applicability.

Adapted for this guideline from: American Academy of Pediatrics Steering Committee on Quality Improvement and Management. Classifying recommendations for clinical practice guidelines. Pediatrics 2004; 114(3):874–7.

Clinical Algorithm(s)

None provided

Scope

Disease/Condition(s)

Phenylketonuria (PKU), including PKU before, during and after pregnancy

Guideline Category

Evaluation

Management

Treatment

Clinical Specialty

Family Practice

Internal Medicine

Medical Genetics

Nutrition

Obstetrics and Gynecology

Pediatrics

Intended Users

Advanced Practice Nurses

Dietitians

Health Care Providers

Physician Assistants

Physicians

Guideline Objective(s)

To establish harmonization in treatment and monitoring, to guide the integration of nutrition therapy in the medical management of phenylketonuria (PKU), and to improve outcomes (nutritional, cognitive, and developmental) for individuals with PKU in all life stages while reducing associated medical, educational, and social costs

Target Population

- Individuals with phenylketonuria (PKU) at all life stages from the newborn period, through childhood and adolescence, to ongoing management in adult years, and during pregnancy
- Individuals who were late-diagnosed or are returning to treatment and those pairing dietary treatment with other treatment modalities

Interventions and Practices Considered

1. Nutrition interventions for appropriate intake
    - Meeting the individual's recommended phenylalanine (PHE) intake
    - Adjusting total protein intake from a combination of intact protein and amino acid-based medical food to age-recommended levels
    - Supplemental tyrosine (TYR)
    - Meeting dietary reference intakes (DRIs) for all other nutrients/micronutrients and energy
2. Maintaining appropriate lifelong blood levels of PHE
3. Strategies for nutrition intervention
    - Choosing medical foods and supplements to meet recommended nutrient intake and maintaining optimal blood PHE levels
    - Encouraging use of breast milk in infants
    - Gradual introduction of solid foods in infants
    - Minimizing elevation of blood PHE during illness
    - Ensuring appropriate intake of PHE by having accurate data about food PHE content and planning and monitoring of dietary PHE intake
    - Encouraging lifelong adherence to nutritional recommendations
4. Monitoring nutrition interventions
    - Monitoring dietary records to assess adequacy of nutrient intake in supporting appropriate growth and nutritional status
    - Monitoring age-specific anthropometrics
    - Routine monitoring of clinical indicators and biochemical markers for deficiency or excess of nutrients (e.g., PHE, TYR, protein, iron and vitamin D)
    - Monitoring clinical indicators and biochemical markers when indicated by special circumstances (e.g., rapid growth, pregnancy, poor compliance)
    - Monitoring neurocognitive development
    - Assessing quality of life
5. Use of adjunctive therapies (sapropterin, large neutral amino acids [LNAA])
6. Nutritional interventions before, during, and after pregnancy

- Maintaining blood PHE levels
- Monitoring weight gain, dietary intake, and biochemical parameters
- Prescribing appropriate diet
- Avoiding LNAA therapy during pregnancy
- Evaluating use of sapropterin during pregnancy on a case-by-case basis
- Facilitating access to psychosocial support
- Encouraging maintenance of dietary therapy after pregnancy and breastfeeding of infants
- Modifying therapy and collaboration with other care-givers under special circumstances

Major Outcomes Considered

- Quality of life
- Nutritional status including prevalence of nutritional deficiencies
- Physiological and neurobehavioral outcomes (e.g., level of cognitive disability, psychiatric symptoms, eczema, and impaired socialization)
- Growth and development
- Blood phenylalanine (PHE) levels and dietary PHE tolerance
- Treatment adherence
- Prevalence of overweight and obesity
- Birth defect incidence and cognitive outcome of offspring
- Fetal growth and development

Methodology

Methods Used to Collect/Select the Evidence

Hand-searches of Published Literature (Primary Sources)

Hand-searches of Published Literature (Secondary Sources)

Searches of Electronic Databases

Searches of Unpublished Data

Description of Methods Used to Collect/Select the Evidence

Search Process

The search process, critical appraisal and abstraction, and consensus input and evidence summary were previously described in the Maple Syrup Urine Disease guidelines and are outlined on the Genetic Metabolic Dietitians International (GMDI)/Southeast Regional Newborn Screening and Genetics Consortium (SERC) portal (https://GMDI.org/). PubMed searches included human studies (excluding genetic studies not related to treatment) published in English from 1980 to late 2000 and from 2011 to February 2014. Studies published from 2000 through 2010 were reviewed and summarized for the National Institutes of Health (NIH) State of the Science Conference (February 2013) by dietitians who were members of the NIH Diet Management Working Group or Maternal PKU Working Group, and who were also involved in this GMDI/SERC guideline effort. The summary statement from the State of the Science Conference is referenced in the guideline in lieu of individual articles used in preparation of the summary statement. Literature reviewed for the published companion American College of Medical Genetics (ACMG) Guidelines and GMDI/SERC Recommendations are similarly referenced.

Gray literature refers to resources that cannot be accessed through standard search systems. These include abstracts and presentations from scientific meetings, clinical protocols and guidelines, unpublished research, communication among experts (including list-serves), professional newsletters, and book chapters. Gray literature related to phenylketonuria (PKU) was collected by work group members and was screened and prioritized for inclusion based on relevance, currency, and substantive information not available in scientific literature.

Formal Literature Search Plan

Question: For individuals with PKU what nutrient intakes are associated with positive outcomes?

- Search Terms: "Phenylketonuria", and "treatment" "diet" "nutrition" "requirements" "trace minerals" "protein" "vitamins", "essential fatty acids" "medical food" "phenylalanine" "tyrosine" "micronutrients" "nutritional status"
- Date of Literature Search Request: October 2, 2012

Question: For individuals with PKU what blood phenylalanine concentration is associated with positive outcomes?

- Search Terms: "Blood phenylalanine" "PKU" AND "outcome" "neuro-cognitive function" "intelligence" "executive function" "development" "behavior" "working memory" "quality of life"
- Date of Literature Search Request: October 2, 2012

Question: For individuals with PKU which nutrition interventions are associated with positive outcomes?

- Search Terms: "Phenylketonuria" and "breast feeding", "counseling", "compliance", "adherence", "self-management" "nutrition education" "quality of life" "low protein foods" "diet" "diet therapy" "dietary control" "management" "weaning" "medical food" "dietary phenylalanine distribution" "infant-newborn" "PKU diet" AND "prematurity", "pregnancy" "maternal PKU" "MPKU" "lactation", "late-treated" "adult" "prevention" "birth defects" "total parenteral nutrition" "custom amino acid solutions" "cost" "resource use" "budget impact" "optimal and consistent care" "follow-up"
- Date of Literature Search Request: October 2, 2012

Question: For individuals with PKU monitoring of which parameters is associated with positive outcomes?

- Search Terms: "Phenylketonuria" AND "nutritional deficits", "nutritional status", "prealbumin" "plasma amino acids" "growth" "obesity" "bone mineral density" "bone age" "monitoring" "development" "physical development" "overweight with PKU" "BMI" "alopecia" "hair loss"
- Date of Literature Search Request: October 2, 2012

Question: For individuals with PKU whose therapy includes adjunctive therapy or other therapy options (sapropterin or large neutral amino acids) what dietary considerations are needed for positive outcomes?

- Search Terms: "Phenylketonuria" and "Kuvan", "BH4", "tetrahydrobiopterin", "sapropterin", "large neutral amino acids", "LNAA", "glycomacropeptides", "GMP" "whey protein"
- Date of Literature Search Request: October 2, 2012

Question: For women with PKU, what nutritional therapies are associated with positive outcomes during pregnancy planning, pregnancy, and the post-partum period (including lactation)?

- Search Terms: "PKU", "Phenylketonuria", "phenylalanine", "MPKU", "maternal PKU" and "treatment", "diet", "pregnancy", "nutritional requirements", "lactation"
- Date of Literature Search Request: November 10, 2012

Number of Source Documents

Of 1596 identified sources that met inclusion criteria, 239 peer-reviewed and 25 gray literature sources were adjudicated to be analyzed and became part of the evidence synthesis (including 40 for nutrient intake, 26 for blood phenylalanine (PHE) concentration, 121 for nutrition intervention, 65 for monitoring, 59 for adjunctive therapies, and 30 for pregnancy). Additionally, a literature synthesis from the National Institutes of Health (NIH) Phenylketonuria Scientific Review Conference was incorporated.

Methods Used to Assess the Quality and Strength of the Evidence

Weighting According to a Rating Scheme (Scheme Not Given)

Rating Scheme for the Strength of the Evidence

Not stated

Methods Used to Analyze the Evidence

Systematic Review with Evidence Tables

Description of the Methods Used to Analyze the Evidence

Critical Appraisal and Abstraction

Each peer-reviewed publication was critically reviewed by a trained analyst using a Quality Criteria Checklist adapted for this purpose. Quality criteria included selection and retention of subjects, appropriate controls, intervention clearly described, other intervening variables tracked, outcomes defined, measures valid and statistical analyses appropriate. Based on the number of criteria met, each article was given a quality rating of positive, neutral or negative. Workgroup members were assigned articles pertinent to a particular research question, which they reviewed and abstracted. Description included: study sample, intervention or factors of interest, findings, limitations of the study, and the article's contribution to the evidence analysis research question.

Workgroup members, using a specially designed tool, subjected eligible gray literature to similar quality assessment and abstraction.

Evidence Summary

Key information from all eligible evidence sources (peer-reviewed and gray literature) for each question was summarized. Summarized evidence was used to draft preliminary conclusion statements and recommendations.

Methods Used to Formulate the Recommendations

Expert Consensus (Delphi)

Expert Consensus (Nominal Group Technique)

Description of Methods Used to Formulate the Recommendations

Question Formulation

Topics for evidence analysis were selected from areas of uncertainty and variation in practice by a phenylketonuria (PKU) working group comprised of ten experienced metabolic dietitians. Six practice-based questions were identified for evidence analysis and guideline development (see Table 1 in the original guideline document). Research questions were formulated in the PICO (population, intervention, comparison, and outcomes) format, and a separate systematic literature search, appraisal, and evidence analysis and summary was completed for each question.

Evidence Summary

Key information from all eligible evidence sources (peer-reviewed and gray literature) for each question was summarized. Summarized evidence was used to draft preliminary conclusion statements and recommendations.

Through two separate Delphi surveys and a nominal group process meeting, clinical expertise was gathered regarding practices not addressed sufficiently in the literature, or for which evidence was inconclusive. The goal was to develop consensus or to note where consensus does not exist. Participants in the Delphi survey were metabolic dietitians and metabolic physicians representing all geographical regions in the U.S. Participants in the nominal group meeting included: a general pediatrician, a researcher, and an advocate for the PKU community, in addition to metabolic physicians and dietitians.

The final conclusion statement for each research question is based on a synthesis of evidence from peer-reviewed publications, gray literature, and Delphi and nominal group consensus techniques.

Guideline Development

The final conclusion statement and recommendations for the nutrition management of individuals with PKU in each of the six topic areas were based on synthesis of all evidence and consensus sources. These were written, reviewed, and edited by the project core group. A guideline consultant, not involved in developing the recommendations, and with input from principal investigators (PIs) and co-PIs with domain expertise, rated each recommendation with respect to strength of the evidence behind the recommendation (strong, fair, limited, consensus, insufficient

evidence) and need for clinical action (imperative [i.e., broadly applicable to individuals with PKU] or conditional [i.e., applicable in specific situations]). Definitions for strength of evidence and clinical action ratings are described in the "Rating Scheme for the Strength of the Recommendations" field.

Rating Scheme for the Strength of the Recommendations

Recommendation Ratings and Clinical Actions/Application

Strength of Evidence for Recommendation Ratings	
Strong	The benefits clearly exceed the harms (or the harms clearly exceed the benefits in the case of a strong negative recommendation); and the quality of the supporting evidence is good. In some clearly identified circumstances, strong recommendations may be made based on lesser evidence when high-quality evidence is impossible to obtain and the anticipated benefits strongly outweigh the harms.
Fair	The benefits exceed the harms (or the harms clearly exceed the benefits in the case of a negative recommendation), but the quality of evidence is not as strong as above. In some clearly identified circumstances, recommendations may be made based on lesser evidence when high-quality evidence is impossible to obtain and the anticipated benefits outweigh the harms.
Weak	The quality of evidence that exists is suspect or well-done studies show little clear advantage for one approach over another.
Consensus	Expert opinion (determined from consensus methodology) supports the recommendation even though the available scientific evidence did not present consistent results, or studies were lacking.
Insufficient Evidence	There is a lack of pertinent evidence (from research and clinical practice) and/or an unclear balance between benefits and harms.
Clinical Action/Application	
Imperative	The recommendation is broadly applicable to the target population without conditions.
Conditional	The recommendation clearly defines a specific situation that limits its applicability.

Adapted for this guideline from: American Academy of Pediatrics Steering Committee on Quality Improvement and Management. Classifying recommendations for clinical practice guidelines. Pediatrics 2004; 114(3):874–7.

Cost Analysis

A formal cost analysis was not performed and published cost analyses were not reviewed.

Method of Guideline Validation

External Peer Review

Description of Method of Guideline Validation

The final document was reviewed by an external panel using the Appraisal of Guidelines for Research and Evaluation (AGREE II). The external panel consisted of three metabolic physicians, three dietitians, and an expert in guideline development and methodology, who were not involved in the evidence analysis nor in the development phases of the phenylketonuria (PKU) guideline.

Evidence Supporting the Recommendations

Type of Evidence Supporting the Recommendations

The type of supporting evidence is specifically stated for each recommendation (see the "Major Recommendations" field).

Benefits/Harms of Implementing the Guideline Recommendations

Potential Benefits

Implementing the recommendations would:

- Reduce variations in clinical practice and services across medical centers
- Guide practice decisions that integrate medical and nutrition management/therapy
- Provide clinicians with criteria to make recommendations for nutrition management or recommend adjuvant therapies
- Design quality nutrition care based on individual metabolic and/or genetic alteration
- Improve clinical outcomes and clinician effectiveness
- Enhance quality of life, prevent untoward consequences and complications, and reduce associated medical, educational and social costs

Potential Harms

This guideline recommends that individuals with phenylketonuria (PKU) maintain blood phenylalanine (PHE) between 120 and 360 µmol/L through their lifetime. Unnecessary treatment, or more stringent treatment than needed for optimal outcomes, has the potential to create psychosocial costs for individuals with PKU. Such treatment may be burdensome and stigmatizing, may have nutritional consequences (vitamin/mineral/protein deficiencies), and may increase health care costs, especially since access to medical foods and care is difficult for many. Many older children and adults currently do not adhere well to more liberal blood PHE recommendations so may be unlikely to attain more strict blood PHE recommendations.

Individuals who do not adhere to all aspects of therapy are also at risk for the consequences of elevated blood PHE and possible nutritional deficiencies.

Side effects of sapropterin, used as an adjuvant therapy, include gastrointestinal distress (especially if the drug is not consumed with food) and headaches. The safety of use during pregnancy and lactation in women with PKU is still being studied.

While large neutral amino acids, used as an adjuvant therapy, have allowed improvement in psychosocial characteristics in some individuals with PKU, they do not result in blood PHE in the target range.

Qualifying Statements

Qualifying Statements

This nutrition management guideline is meant to serve as a framework for providing nutrition care to individuals with phenylketonuria (PKU). It may not always be appropriate to use these guidelines. Individual circumstances and complicating conditions must be taken into consideration. The clinical judgment of the health care provider and patient preferences and values dictate treatment decisions. These nutrition management guidelines are provided with the express understanding that they do not establish or specify particular standards of care, whether legal, medical, or other.

Implementation of the Guideline

Description of Implementation Strategy

An implementation strategy was not provided.

Implementation Tools

Patient Resources

Resources

Tool Kits

For information about availability, see the *Availability of Companion Documents* and *Patient Resources* fields below.

Institute of Medicine (IOM) National Healthcare Quality Report Categories

IOM Care Need

Living with Illness

Staying Healthy

IOM Domain

Effectiveness

Patient-centeredness

Identifying Information and Availability

Bibliographic Source(s)

Singh RH, Cunningham AC, Mofidi S, Douglas TD, Frazier DM, Hook DG, Jeffers L, McCune H, Moseley KD, Ogata B, Pendyal S, Skrabal J, Splett PL, Stembridge A, Wessel A, Rohr F. Updated, web-based nutrition management guideline for PKU: an evidence and consensus based approach. Mol Genet Metab. 2016 Jun;118(2):72-83. [118 references] PubMed

Adaptation

Not applicable: The guideline was not adapted from another source.

Date Released

2016 Jun

Guideline Developer(s)

Genetic Metabolic Dietitians International & Southeast Regional Newborn Screening and Genetics Consortium - Independent Expert Panel

Source(s) of Funding

Partial funding for this project has been provided to Southeast Regional Newborn Screening and Genetics Consortium (SERC) by the Health Resources and Services Administration (HRSA): grant # H46MC24090. Genetic Metabolic Dietitians International (GMDI) has received unrestricted education grants from National Phenylketonuria (PKU) Alliance and from Biomarin Pharmaceutical, Inc.

Guideline Committee

PKU Working Group

Composition of Group That Authored the Guideline

Authors: Rani H. Singh (*Nutrition Guidelines Project Principal Investigator*), Department of Human Genetics, Emory University School of Medicine, Atlanta, GA, USA, Nutrition Health Sciences Program, Graduate Division of Biological and Biomedical Sciences, Emory University School of Arts and Sciences, Atlanta, GA, USA; Amy C. Cunningham (*Co-Chair of PKU Working Group*), Hayward Genetics Center, Tulane University School of Medicine, New Orleans, LA, USA; Shideh Mofidi (*Co-Chair of PKU Working Group*), Inherited Metabolic Disease Center, Maria Fareri Childrens Hospital, Westchester Medical Center, New York Medical College, Valhalla, New York, USA; Teresa D. Douglas, Department of Human Genetics, Emory University School of Medicine, Atlanta, GA, USA; Dianne M. Frazier, Division of Genetics and Metabolism, University of North Carolina School of Medicine, Chapel Hill, NC, USA; Debra Geary Hook, University of California, Davis, USA; Laura Jeffers, Cleveland Clinic, Center for Human Nutrition, Cleveland, OH, USA; Helen McCune, Pediatric Genetics and Metabolism, University of Florida, Gainesville, FL, USA; Kathryn D. Moseley, Genetics Division, USC/Keck School of Medicine, Los Angeles, CA, USA; Beth Ogata, University of Washington, Department of Pediatrics, Seattle, WA, USA; Surekha Pendyal, Division of Genetics and Metabolism, University of North Carolina School of Medicine, Chapel Hill, NC, USA; Jill Skrabal, Department of Medical Genetics, University of Nebraska Medical Center/Children's Hospital and Medical Center, Nebraska Medical Center, Omaha, NE, USA; Patricia L. Splett, Evaluation Consultant, Splett & Associates, LLC, Stanchfield, MN, USA; Adrya Stembridge, Department of Human Genetics, Emory University School of Medicine, Atlanta, GA, USA; Ann Wessel, Division of Genetics and Genomics, Boston Children's Hospital, Boston, MA, USA; Frances Rohr (*Nutrition Guidelines Project Principal Investigator*), Division of Genetics and Genomics, Boston Children's Hospital, Boston, MA, USA

Financial Disclosures/Conflicts of Interest

Conflict of Interest Statement

- Rani H. Singh, PhD, RD has served on the global medical advisory board for Nutricia and has received payment.
- Amy Cunningham has received compensation for faculty participation at Metabolic University, which is supported by Nutricia, and a stipend for speaking at the Abbott Nutrition Metabolic Conference. She participates on advisory boards for BioMarin Pharmaceutical, for which she receives travel funding and stipend support. She functions as project coordinator of research studies for which the Hayward Genetics Center receives grant funding from BioMarin Pharmaceutical.
- Shideh Mofidi has no financial relationships relevant to this article to disclose.
- Teresa Douglas has no financial relationships relevant to this article to disclose.
- Dianne Frazier has received speaker/travel funds from Abbott Nutrition, an honorarium from the Nutricia Advisory Board and an educational grant from Genetic Metabolic Dietitians International.
- Debra Hook has participated consulting work for Hyperion, BioMarin, and Ultragenyx, as well as speaking and projects for Vitaflo, Nutricia, and Cambrooke.
- Laura Jeffers has served on the speakers Bureau for BioMarin.
- Helen McCune has received honorarium from the BioMarin Advisory Board.
- Kathryn Moseley has no financial relationships relevant to this article to disclose.
- Beth Ogata has no financial relationships relevant to this article to disclose.
- Surekha Pendyal has no financial relationships relevant to this article to disclose.
- Jill Skrabal has a consulting agreement with Nutricia and BioMarin.
- Patricia L. Splett has no financial relationships relevant to this article to disclose.
- Adrya Stembridge has no financial relationships relevant to this article to disclose.
- Ann Wessel has no financial relationships relevant to this article to disclose.
- Fran Rohr has received consulting fees and/or travel stipend from Abbott, BioMarin, Nutricia, Vitaflo USA.

Guideline Status

This is the current release of the guideline.

This guideline meets NGC's 2013 (revised) inclusion criteria.

Guideline Availability

Available to subscribers from the Molecular Genetics and Metabolism Web site .

Availability of Companion Documents

The following are available:

- The HTML version of the guideline, with additional tables and other resources, is available from the Southeast Regional Newborn Screening and Genetics Collaborative Web site .
- A PKU Nutrition Management Toolkit is available from the Southeast Regional Newborn Screening and Genetics Collaborative Web site .

Patient Resources

A compilation of resources for patients and their families is available from the National PKU Alliance Web site .

Please note: This patient information is intended to provide health professionals with information to share with their patients to help them better understand their health and their diagnosed disorders. By providing access to this patient information, it is not the intention of NGC to provide specific medical advice for particular patients. Rather we urge patients and their representatives to review this material and then to consult with a licensed health professional for evaluation of treatment options suitable for them as well as for diagnosis and answers to their personal medical questions. This patient information has been derived and prepared from a guideline for health care professionals included on NGC by the authors or publishers of that original guideline. The patient information is not reviewed by NGC to establish whether or not it accurately reflects the original guideline's content.

NGC Status

This NGC summary was completed by ECRI Institute on January 5, 2017. The information was not verified by the guideline developer.

Copyright Statement

Disclaimer

NGC Disclaimer